



Single Nucleotide Polymorphism



PubMed Nucleotide Protein Genome Structure PopSet Taxonomy

Search for

[Limits](#)[Preview](#)[Index](#)[History](#)[Clipboard](#)[Details](#)

Reference SNP Cluster Report

GENERAL

Contact Us
dbSNP Homepage
SNP Science Primer
Announcements
dbSNP Summary
FTP SERVER
Getting Started
Build History
Handle Request

NCBI SNP CLUSTER ID: **rs7592**
Organism: human (*Homo sapiens*)
Variation Class: SNP: single nucleotide polymorphism
Molecule Type: Genomic
dbSNP build of first appearance: 52
dbSNP build of most recent change to cluster: 116

SNP Details are categorized in the following sections:

[Submission](#) [Fasta](#) [Resource](#) [Locus](#) [Map](#) [Variation](#) [Va](#)

DOCUMENTATION

FAQ
Overview
How to Submit
RefSNP Summary Info
Database Schema
pdf
Changes **NEW**
Data Formats
Heterozygosity
Computation

SEARCH

Entrez SNP
Blast SNP
Batch Query
By Submitter
New Batches
Method
Population
Detail
Class
Publication
Chromosome Report
Locus Information
STS Markers
Free Form Search
Simple
Advanced

HAPLOTYPE

Specifications
Sample HapSet
Sample Individual

Submitter records for this RefSNP Cluster

The submission **ss11944722** has the longest flanking sequence of all cluster members and was us current build.

NCBI Assay ID	Handle Submitter ID	Validation Status	Entry Date	Update Date
ss9489	CGAP-GAI 50173		08/23/99	01/29/0
ss20257	UWGC 2197		11/15/99	01/29/0
ss20261	UWGC 2201		11/15/99	01/29/0
ss1510338	LEE 34488		09/13/00	01/04/0
ss1510475	LEE 38702		09/13/00	01/04/0
ss3194964	YUSUKE IMS-JST010882		08/07/01	01/04/0
ss4404192	LEE e34488		04/26/02	07/03/0
ss4404281	LEE e38702		04/26/02	07/03/0
ss11944722	WI_SSAHASNP chr8.NT_023666.15_396196		07/04/03	07/04/0

Fasta sequence (**Legend**)

>gnl|dbSNP|rs7592|allelePos=201|totalLen=401|taxid=9606|snpclass=1|alleles='A/G'|mol=Genom

```
TCATCCAGGC AACTCGGGGG AGGGGAAGCT CACAGACCGG TACTTCCCAC TCCCCTGATT
CTCTCTGTCC ATCCTCAACA TTCCTTTGCT TCACAGGGTC AGTGAAGCC CCAACGGGAA
AGGAAACGCC CCGGGCAAAG GGTCTTTTGC AGCTTTTGCA GACGGGCAAG AAGCTGCTTC
TGCCACACC GCAGGGACAA
R
```

NCBI Resource Links

dbSTS:

GenBank mRNA: AK058094.1 BC005913.1 J03517.1 J03553.1

UniGene Cluster ID: 1074

8 [NT_023666.15](#) 396196 21842452 plus strand

NCBI Sequence Viewer: See [rs7592](#) in Sequence Viewer.


Project Ensembl: Query [rs7592](#) in Ensembl.

UC Santa Cruz Genome Assembly: Query [rs7592](#) on the Santa Cruz Assembly.

Variation Summary:

Assay sample size (number of chromosomes):	101
Population data sample size (number of chromosomes):	188
Total number of populations with frequency data:	1
Total number of individuals with genotype data:	0
Average estimated <u>heterozygosity</u> :	0.488
Average Allele Frequency:	
A	0.576
G	0.424

Validation Summary:

Validation status: 

Marker displays Mendelian segregation:	UNKNOWN
PCR results confirmed in multiple reactions:	UNKNOWN
Homozygotes detected in individual genotype data:	UNKNOWN

GENERAL: [Contact Us](#) | [Homepage](#) | [Announcements](#) | [dbSNP Summary](#) | [Genome](#) | [FTP SERVER](#) | [Build History](#) | [Handle Request](#)

DOCUMENTATION: [FAQ](#) | [Overview](#) | [How to Submit](#) | [RefSNP Summary](#) | [Info](#) | [Database Schema](#)

SEARCH: [Entrez SNP](#) | [Blast SNP](#) | [Main Search](#) | [Batch Query](#) | [By Submitter](#) | [New Batches](#) | [Method](#) | [Population](#) | [Publication](#) | [Chromosome Report](#) | [Batch](#) | [Locus Info](#) | [Free Form](#) | [Easy Form](#) | [Between Marker](#)

HAPLOTYPE: [Specifications](#) | [Sample HapSet](#) | [Sample Individual](#)
NCBI: [PubMed](#) | [Entrez](#) | [BLAST](#) | [OMIM](#) | [Taxonomy](#) | [Structure](#)

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Revised August 11, 2003 2:18 PM



Single Nucleotide Polymorphism



[PubMed](#)
[Nucleotide](#)
[Protein](#)
[Genome](#)
[Structure](#)
[PopSet](#)
[Taxonomy](#)

Search for

[Limits](#)
[Preview](#)
[Index](#)
[History](#)
[Clipboard](#)
[Details](#)

Reference SNP Cluster Report

GENERAL

[Contact Us](#)
[dbSNP Homepage](#)
[SNP Science Primer](#)
[Announcements](#)
[dbSNP Summary](#)
[FTP SERVER](#)
[Getting Started](#)
[Build History](#)
[Handle Request](#)

NCBI SNP CLUSTER ID: rs1139547
Organism: human (*Homo sapiens*)
Variation Class: SNP: single nucleotide polymorphism
Molecule Type: cDNA
dbSNP build of first appearance: 86
dbSNP build of most recent change to cluster: 103

SNP Details are categorized in the following sections:

[Submission](#)
[Fasta](#)
[Resource](#)
[Locus](#)
[Map](#)
[Variation](#)
[Va](#)

DOCUMENTATION

[FAQ](#)
[Overview](#)
[How to Submit](#)
[RefSNP Summary Info](#)
[Database Schema](#)
[pdf](#)
[Changes](#) NEW
[Data Formats](#)
[Heterozygosity](#)
[Computation](#)

Submitter records for this RefSNP Cluster

The submission **ss4322597** has the longest flanking sequence of all cluster members and was used during BLAST analysis for the current build.

NCBI Assay ID	Handle Submitter ID	Validation Status	Entry Date	Update Date	Build Added	Molecule Type	Sequen Ori ntat
ss1548788	LEE 1015779		09/13/00	01/29/01	86	cDNA	forward
ss4322597	CGAP- GAI 263851		11/28/01	03/26/02	103	cDNA	reverse

SEARCH

[Entrez SNP](#)
[Blast SNP](#)
[Batch Query](#)
[By Submitter](#)
[New Batches](#)
[Method](#)
[Population](#)
[Detail](#)
[Class](#)
[Publication](#)
[Chromosome Report](#)
[Locus Information](#)
[STS Markers](#)
[Free Form Search](#)
[Simple](#)
[Advanced](#)

Fasta sequence (Legend)

>gnl|dbSNP|rs1139547|allelePos=51|totalLen=107|taxid=9606|snpclass=1|alleles='A/T/G'|mol=cDN

```

CCCTGGAG
AAATGGGAGC TTGGGGAGAG GATGGGAGTG GGCAGAGGTG GC
D
CCCAGGGGCC CGGGAAC TCC TGCCACAACA GAATAAGCA GCCTGATTGA AAAGCA
  
```

NCBI Resource Links

Submitter-Referenced Accessions:

dbSTS:
 GenBank: [AI692500](#) [Hs.1074](#)

dbSNP Blast Analysis:

NCBI RefSeq NM (mRNA): [NM_003018.1](#)
 GenBank STS: [G62003.1](#) [G62006.1](#)
 GenBank mRNA: [AK058094.1](#) [BC005913.1](#) [J03517.1](#) [J03553.1](#)

HAPLOTYPE

[Specifications](#)
[Sample HapSet](#)
[Sample Individual](#)

LocusLink Analysis

LocusLink via analysis of contig annotation: [BMP1](#) bone morphogenetic protein 1

Variation has been mapped within 2 kb of a mRNA transcript for bone morphogenetic protein 1

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396247		locus				

LocusLink via analysis of contig annotation: [SFTPC](#) surfactant, pulmonary-associated protein C

Gene Model (contig mRNA transcript) information from genome sequence for [NM_003018](#)



Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396247		untranslated region				

LocusLink: no link established by BLAST analysis of mRNA sequences

Integrated Maps:

NCBI MapViewer: [rs1139547](#) maps exactly once on NCBI human [chromosome 8](#)

Chromosome	Contig accession	Contig Position	Chromosome Position	Hit orientation
8	NT_023666.15	396247	21842503	plus strand

NCBI Sequence Viewer: See [rs1139547](#) in Sequence Viewer.

Project Ensembl: Query [rs1139547](#) in Ensembl.

UC Santa Cruz Genome Assembly: Query [rs1139547](#) on the Santa Cruz Assembly.

Variation Summary:

Assay sample size (number of chromosomes):	31
Population data sample size (number of chromosomes):	
Total number of populations with frequency data:	0
Total number of individuals with genotype data:	0
Average estimated heterozygosity:	Not available

Validation Summary:

Validation status:

Marker displays Mendelian segregation:	UNKNOWN
PCR results confirmed in multiple reactions:	UNKNOWN
Homozygotes detected in individual genotype data:	UNKNOWN

GENERAL: [Contact Us](#) | [Homepage](#) | [Announcements](#) | [dbSNP Summary](#) | [Genome](#) | [FTP SERVER](#) | [Build History](#) | [Handle Request](#)

DOCUMENTATION: [FAQ](#) | [Overview](#) | [How to Submit](#) | [RefSNP Summary Info](#) | [Database Schema](#)

SEARCH: [Entrez SNP](#) | [Blast SNP](#) | [Main Search](#) | [Batch Query](#) | [By Submitter](#) | [New Batches](#) | [Method](#) | [Population](#) | [Publication](#) | [Chromosome Report](#) | [Batch](#) | [Locus Info](#) | [Free Form](#) | [Easy Form](#) | [Between Marker](#)

HAPLOTYPE: [Specifications](#) | [Sample HapSet](#) | [Sample Individual](#)

NCBI: [PubMed](#) | [Entrez](#) | [BLAST](#) | [OMIM](#) | [Taxonomy](#) | [Structure](#)

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Revised August 11, 2003 2:18 PM



Single Nucleotide Polymorphism



[PubMed](#)
[Nucleotide](#)
[Protein](#)
[Genome](#)
[Structure](#)
[PopSet](#)
[Taxonomy](#)

Search for

[Limits](#)
[Preview](#)
[Index](#)
[History](#)
[Clipboard](#)
[Details](#)

Reference SNP Cluster Report

GENERAL

[Contact Us](#)
[dbSNP Homepage](#)
[SNP Science Primer](#)
[Announcements](#)
[dbSNP Summary](#)
[FTP SERVER](#)
[Getting Started](#)
[Build History](#)
[Handle Request](#)

NCBI SNP CLUSTER ID: rs1126931
Organism: human (*Homo sapiens*)
Variation Class: SNP: single nucleotide polymorphism
Molecule Type: cDNA
dbSNP build of first appearance: 86
dbSNP build of most recent change to cluster: 103

SNP Details are categorized in the following sections:

[Submission](#)
[Fasta](#)
[Resource](#)
[Locus](#)
[Map](#)
[Variation](#)
[Va](#)

DOCUMENTATION

[FAQ](#)
[Overview](#)
[How to Submit](#)
[RefSNP Summary Info](#)
[Database Schema](#)
[pdf](#)
[Changes](#) NEW
[Data Formats](#)
[Heterozygosity](#)
[Computation](#)

Submitter records for this RefSNP Cluster

The submission **ss1510417** has the longest flanking sequence of all cluster members and was used during BLAST analysis for the current build.

NCBI Assay ID	Handle	Submitter ID	Validation Status	Entry Date	Update Date	Build Added	Molecule Type	Sequence Orientation
ss1510417	LEE	36582		09/13/00	01/29/01	86	cDNA	forward
ss4322596	CGAP-	GAI263848		11/28/01	03/26/02	103	cDNA	reverse

SEARCH

[Entrez SNP](#)
[Blast SNP](#)
[Batch Query](#)
[By Submitter](#)
[New Batches](#)
[Method](#)
[Population](#)
[Detail](#)
[Class](#)
[Publication](#)
[Chromosome Report](#)
[Locus Information](#)
[STS Markers](#)
[Free Form Search](#)
[Simple](#)
[Advanced](#)

Fasta sequence (Legend)

>gnl|dbSNP|rs1126931|allelePos=51|totalLen=101|taxid=9606|snpclass=1|alleles='C/T'|mol=cDNA

```

ACCCTGGAGA AATGGGAGCT TGGGGAGAGG ATGGGAGTGG GCAGAGGTGG
Y
ACCCAGGGGC CCGGGAATC CTGCCACAAC AGAATAAAGC AGCCTGATTG
  
```

NCBI Resource Links

Submitter-Referenced Accessions:

dbSTS:
 GenBank: [AI203517](#) [Hs.1074](#)

dbSNP Blast Analysis:

NCBI RefSeq NM (mRNA): [NM_003018.1](#)
 GenBank HTGS Finished: [AC105141.7](#) [AC105206.8](#) [J03890.1](#) [U02948.1](#)
 GenBank STS: [G62003.1](#) [G62006.1](#)
 GenBank mRNA: [AK058094.1](#) [BC005913.1](#) [J03517.1](#) [J03553.1](#)

UniGene transcribed sequence cluster:

HAPLOTYPE

[Specifications](#)
[Sample HapSet](#)
[Sample Individual](#)

UniGene Cluster ID: [1074](#)**LocusLink Analysis****LocusLink** via analysis of contig annotation: [BMP1](#) bone morphogenetic protein 1

Variation has been mapped within 2 kb of a mRNA transcript for bone morphogenetic protein 1

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396246		locus				

LocusLink via analysis of contig annotation: [SFTPC](#) surfactant, pulmonary-associated protein CGene Model (contig mRNA transcript) information from genome sequence for [NM_003018](#)

Contig accession	Contig position	Protein accession	Function	dbSNP allele	Protein residue	Codon position	Amino acid position
NT_023666	396246		untranslated region				

LocusLink via BLAST analysis of mRNAs: [SFTPC](#) surfactant, pulmonary-associated protein C
Variations are assigned to a gene if mapped within 2 kb of mRNA sequence feature.

Accession class	Nucleotide accession	Nucleotide Position	Hit orientation	Protein accession	Function
NCBI RefSeq	NM_003018.1	794	plus strand	NP_003009.1	unclassified
GenBank mRNA	BC005913.1	790	plus strand	AAH05913.1	unclassified
GenBank mRNA	J03517.1	794	plus strand	AAA36634.1	unclassified
GenBank mRNA	J03553.1	941	plus strand	AAA36631.1	unclassified

Integrated Maps:**NCBI MapViewer:** [rs1126931](#) maps exactly once on NCBI human [chromosome 8](#)

Chromosome	Contig accession	Contig Position	Chromosome Position	Hit orientation
8	NT_023666.15	396246	21842502	plus strand

NCBI Sequence Viewer: See [rs1126931](#) in Sequence Viewer.**Project Ensembl:** Query [rs1126931](#) in Ensembl.**UC Santa Cruz Genome Assembly:** Query [rs1126931](#) on the Santa Cruz Assembly.**Variation Summary:**

Assay sample size (number of chromosomes):	36
Population data sample size (number of chromosomes):	
Total number of populations with frequency data:	0
Total number of individuals with genotype data:	0
Average estimated heterozygosity :	Not available

Validation Summary:**Validation status:**

Marker displays Mendelian segregation: UNKNOWN

PCR results confirmed in multiple reactions: UNKNOWN

Homozygotes detected in individual genotype data: UNKNOWN

GENERAL: [Contact Us](#) | [Homepage](#) | [Announcements](#) | [dbSNP Summary](#) | [Genome](#) | [FTP SERVER](#) | [Build History](#) | [Handle Request](#)**DOCUMENTATION:** [FAQ](#) | [Overview](#) | [How to Submit](#) | [RefSNP Summary](#) | [Info](#) | [Database Schema](#)**SEARCH:** [Entrez SNP](#) | [Blast SNP](#) | [Main Search](#) | [Batch Query](#) | [By Submitter](#) | [New Batches](#) | [Method](#) | [Population](#) | [Publication](#) | [Chromosome Report](#) | [Batch](#) | [Locus Info](#) | [Free Form](#) | [Easy Form](#) | [Between Marker](#)**HAPLOTYPE:** [Specifications](#) | [Sample HapSet](#) | [Sample Individual](#)**NCBI:** [PubMed](#) | [Entrez](#) | [BLAST](#) | [OMIM](#) | [Taxonomy](#) | [Structure](#)[Disclaimer](#) | [Privacy statement](#)

Revised August 11, 2003 2:18 PM

FILE 'MEDLINE' ENTERED AT 14:17:35 ON 10 SEP 2003
ACTIVATE SURFACTANT/L

L1 (1623) SEA PLU=ON SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W) (C OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A) (C OR 2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A) (2 OR C)) OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN SP5)

L2 (2417) SEA PLU=ON SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W) (C OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A) (C OR 2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A) (2 OR C)) OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN SP5)

L3 (4226) SEA PLU=ON SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W) (C OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A) (C OR 2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A) (2 OR C)) OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN SP5)

L4 (8266) SEA PLU=ON SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W) (C OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A) (C OR 2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A) (2 OR C)) OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN SP5)

L5 (603) SEA PLU=ON L1 AND SURFACTANT

L6 (722) SEA PLU=ON L2 AND SURFACTANT

L7 (846) SEA PLU=ON L3 AND SURFACTANT

L8 (2171) SEA PLU=ON L4 AND SURFACTANT

L9 (18) SEA PLU=ON L5 AND (L1(8A) (SNP OR POLYMORPH? OR ALLEL? OR MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))

L10 (26) SEA PLU=ON L6 AND (L2(8A) (SNP OR POLYMORPH? OR ALLEL? OR MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))

L11 (18) SEA PLU=ON L7 AND (L3(8A) (SNP OR POLYMORPH? OR ALLEL? OR MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))

L12 (62) SEA PLU=ON L8 AND (L4(8A) (SNP OR POLYMORPH? OR ALLEL? OR MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))

L13 (37) DUP REM L12 (25 DUPLICATES REMOVED)

FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 14:18:25 ON 10 SEP 2003

L14 44 SEA PLU=ON L12

L15 8265 SEA PLU=ON SFTPC OR SFTP2 OR SP-C OR SP5 OR SPC OR (SFTP(W) (C OR 2)) OR (SURFACTANT(3A) PULMONARY(3A) ASSOCIATED(3A) (C OR 2)) OR (PULMONARY(3A) SURFACTANT(3A) APOPROTEIN(5A) (2 OR C)) OR (SURFACTANT(3A) PROTEOLIPID) OR (PULMONARY SURFACTANT PROTEIN SP5)

L16 2171 SEA PLU=ON L15 AND SURFACTANT

L17 62 SEA PLU=ON L16 AND (L1(8A) (SNP OR POLYMORPH? OR ALLEL? OR MUTAT? OR VARIAN? OR RFLP OR RESTRICTION))

L18 37 DUP REM L17 (25 DUPLICATES REMOVED)

D TI 1-37

D IBIB AB 35

D KWIC 35

D IBIB AB 27

D IBIB AB 2-4 8 11 14 15 17 18 20

FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 15:00:57 ON 10 SEP 2003

L22 24 SEA PLU=ON NOGEE-L/AU

E NOGEE-L/AU

E NOGEE L/AU

L23 127 SEA PLU=ON ("NOGEE L"/AU OR "NOGEE L M"/AU OR "NOGEE LARRY"/AU OR "NOGEE LAWRENCE"/AU OR "NOGEE LAWRENCE M"/AU)

E COLE F/AU

E WHITSETT J/AU

L24	0	SEA PLU=ON	"WHITSETT J"/AU "WHITSETT J A"/AU
		E	WHITSETT J/AU
L25	647	SEA PLU=ON	("WHITSETT J"/AU OR "WHITSETT J A"/AU)
		E	WHITSETT JE/AU
L26	597	SEA PLU=ON	("WHITSETT JE"/AU OR "WHITSETT JEFF"/AU OR
			"WHITSETT JEFF A"/AU OR "WHITSETT JEFFERY A"/AU OR "WHITSETT
			JEFFREY A"/AU OR "WHITSETT JEFFREY"/AU OR "WHITSETT JEFFREY
			A"/AU)
L27	1324	SEA PLU=ON	L26 OR L25 OR L23
L28	987	SEA PLU=ON	L27 AND SURFACTANT
		D	KWIC 1-5
L29	320	SEA PLU=ON	L28 AND L16
L30	305	SEA PLU=ON	L29 NOT L18
L31	303	SEA PLU=ON	L29 NOT L17
L32	13	SEA PLU=ON	L31 AND INTERSTITIAL
L33	7	DUP REM L32	(6 DUPLICATES REMOVED)
		D	TI 1-7
		D	IBIB AB 3

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OM nucleic - nucleic search, using sw model

Run on: September 16, 2003, 13:07:51 ; Search time 2680 Seconds
(without alignments)
351.090 Million cell updates/sec

Title: US-10-074-247-1-C-AT-243
Perfect score: 23
Sequence: 1 ggagatgagcactggggcgccgg 23

Scoring table: OLIGO_NUC
Gapop 60.0 , Gapext 60.0

Searched: 2888711 seqs, 20454813386 residues

Word size : 12

Total number of hits satisfying chosen parameters: 21885

Minimum DB seq length: 0
Maximum DB seq length: 2000000000

Post-processing: Listing first 500 summaries

Database : GenEmbl:*
identical search repeated in GENSEQ, USPATS, USPGPUBS, and EST databases